

TSC Alliance - Mission: The TSC Alliance improves quality of life for everyone affected by tuberous sclerosis complex by catalyzing new treatments, driving research toward a cure and expanding access to lifelong support.

Approved by TSC Alliance Board of Directors: October 13, 2023

| From Where (Referral Sources) | For Whom (Populations, listed in order of life stage) | Assumptions (Theory of Change) | Strategies/Activities (Program Interventions) | Leading Indicators | Outcomes | Impact |
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| <ul style="list-style-type: none"> Parents / Caregivers / Individuals Community Volunteers / Walks and Events TSC Clinics TSC Alliance Website Search Engines Social Media International TSC Groups CDC, NIH Websites Other Advocacy and Umbrella Organizations (e.g., The LAM Foundation, NORD, Global Genes, CNF, REN) Professional Conferences Industry Websites / Outreach Corporate Sponsors Clinicians, Care Providers and Hospitals Case Managers of Government Benefits | <ol style="list-style-type: none"> Prospective parents of those suspected to have TSC in utero Parents/caregivers of infants and children (under 18): <ol style="list-style-type: none"> Diagnosed with TSC Undiagnosed Young adults with TSC (18-26) <ol style="list-style-type: none"> Newly diagnosed young adults Semi-independent and independent adults diagnosed with TSC and their family members Parents/caregivers/ family members of dependent adults diagnosed with TSC Undiagnosed Future parents Adults (27-49) <ol style="list-style-type: none"> Newly diagnosed adults Semi-independent and independent adults diagnosed with TSC and their family members Parents/caregivers/ family members of dependent adults diagnosed with TSC Providers at care facilities Undiagnosed Future parents Older adults (50+) <ol style="list-style-type: none"> Newly diagnosed adults | <p>Our vision: To create a future where everyone affected by tuberous sclerosis complex can live their fullest lives.</p> <p>Tuberous sclerosis complex (TSC) is a rare genetic disease that affects people at all stages of life. TSC causes tumors to grow on different organs, primarily the brain, heart, kidneys, skin, eyes and lungs and can impair their function, TSC can also cause epilepsy; cognitive, behavioral and psychiatric disabilities; aggressive behaviors; and more.</p> <p>Every individual's experience with TSC is different—many are able to live independently despite the disease, while others experience more challenges requiring complex care.</p> <p>TSC is a linchpin disease, meaning its genetic pathway also plays a role in other diseases and disorders. Advancements in TSC research may lead to a better understanding of, autism, epilepsy, traumatic brain injury, diabetes and cancer—diseases that affect more than 65 million people in the US alone.</p> <p>The TSC Alliance® improves quality of life for everyone affected by tuberous sclerosis complex</p> | <p>Accelerate Research</p> <p>Better understand and stratify disease:</p> <ul style="list-style-type: none"> Enable researchers to access and utilize the wide variety of clinical, omics, and Patient Reported Outcomes (PRO) data collected by or shared with TSC Alliance. Increase the number and diversity of individuals with TSC participating in research. Expand quantitative clinical data collected in the Natural History Database (NHD) / Biosample Repository (BSR). Develop prognostic, predictive and stratification biomarkers. Drive development of quantitative patient-reported outcome measures for impactful aspects of TSC. | <p>Accelerate Research</p> <p>Better understand and stratify disease:</p> <ul style="list-style-type: none"> Implement a centralized data library researchers can access (2025). Provide tools for researchers to work with the library of data by implementing a virtual “sandbox” (2028). Ensure at least 10% Black or African American and 4.5% Asian representation in NHD and BSR (US population is 13.6% and 6.1%, respectively). <ul style="list-style-type: none"> From the baseline of 5.9% Black or African American and 2.8% Asian in 2023. Collect quantitative data related to TAND, reproductive and perinatal health. Ensure BSR contains serial blood samples from at least 250 individuals with TSC and access to at least 100 typically developing, non-TSC individuals and at least 100 individuals with TSC but without neurological signs or symptoms. Establish biomarker panel for most impactful manifestations of TSC utilizing samples in BSR and data from clinical studies and NHD. Through external collaborations, generate necessary preliminary data (2025) and pilot newborn screening assay (2028). Establish TAND PRO instrument to quantitatively assess the most impactful TAND symptoms for an individual by 2025 and validate in a clinical study by 2028. | <p>Short-Term Research Outcomes Within 5 years:</p> <ul style="list-style-type: none"> Improved ability to stratify individuals based on risk of severity and types of manifestations to inform clinical treatment and clinical trial design and execution. Increased diversity of investigational tools (e.g., drugs, medical devices, diagnostics) entering clinical trials. <p>Long-Term Research Outcomes Within 5 to 10 years:</p> <ul style="list-style-type: none"> Implementation of precision medicine in the treatment, prevention and development of new therapies for TSC. Advancement of neonatal screening to enable early interventions and preventative care. | <p>Advance treatments to improve clinical outcomes and quality of life for those affected by TSC and alter the course of the disease for future generations.</p> |
| | | | <p>Increase diversity of technologies entering clinical trials:</p> <ul style="list-style-type: none"> Cultivate and engage industry partners to use the Preclinical and Clinical Research Consortia. Enhance collaboration with academic researchers by raising funds to pay for preclinical testing of nominated compounds or other technologies (e.g., gene therapy). Transfer TSC Clinical Research Consortium to the TSC Alliance and establish membership model for industry participation and revenue generation. | <p>Increase diversity of technologies entering clinical trials:</p> <ul style="list-style-type: none"> Maintain at least seven industry partners per year participating in the Preclinical and Clinical Consortia. Test at least 20 compounds or other therapeutic technologies paid for by TSC Alliance in preclinical models including exposure and biomarker response. Establish and make available cell-based neurology model and assay(s) suitable for drug screening. Extend participation in Clinical Research Consortium to all TSC clinics in the U.S., attracting at least two industry or government-funded clinical trials to utilize the Clinical Research Consortium. | | |
| | | | <p>Increase innovative research:</p> <ul style="list-style-type: none"> Advocate for federal and state funding for research. | <p>Increase innovative research:</p> <ul style="list-style-type: none"> Increase TSCRP funding from \$8 million to \$10 million annually. | | |

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| | <p>b. Semi-independent and independent adults diagnosed with TSC and their family members</p> <p>c. Aging parents and caregivers/ family members of dependent adults diagnosed with TSC</p> <p>d. Providers at care facilities</p> <p>e. Undiagnosed</p> <p>6. Parents, caregivers, family members and loved ones of those who have been lost to TSC.</p> | <p>by catalyzing new treatments, driving research toward a cure and expanding access to lifelong support.</p> | <ul style="list-style-type: none"> Expand racial and ethnic diversity of research grant applicants. Demonstrate impact of TSC Alliance funding. | <ul style="list-style-type: none"> Obtain state funding for five TSC centers (currently three states funding four clinics). Fund a minimum of three merit-based research grants per year while growing the diversity of applicants to an average of 15% of applicants from underrepresented populations over five years. Annually capture and report outcomes from funded grants (e.g., follow-on funding, promotion, publications, patents, etc.). | | |
| | | | <p>Improve Access and Quality of Care</p> <p>Expand healthcare access:</p> <ul style="list-style-type: none"> Refine and launch a revamped Professional Advisory Board (PAB), composed of clinicians from diverse specialties serving individuals across the age spectrum and constituent representatives, who respond to and make recommendations to meet the evolving medical needs of individuals and families affected by TSC (e.g., clinical care barriers, clinical trial recruitment/enrollment/equitable representation, changes in health policy). Implement solutions for barriers to care (e.g., decrease clinical trial enrollment time). Expand access to FDA-approved TSC therapies by: 1) identifying states with greatest gaps in insurance coverage requirements for FDA therapies, 2) working with industry leaders and legislators to advocate for change in access criteria in states with greatest gaps, and 3) educating and providing resources to TSC Clinics, Centers of Excellence (COEs) and health care professionals to address denials. Educate the community about which clinics offer telehealth across state lines and to which states they offer it. | <p>Improve Access and Quality of Care</p> <p>Expand healthcare access:</p> <ul style="list-style-type: none"> Develop framework for the PAB for review and approval by the Board of Directors in 2024. <ul style="list-style-type: none"> Hold two meetings per year, beginning in 2025, to identify issues impacting access to care and treatments and potential solutions. In 2024, survey recognized TSC Clinics, COEs and affiliated providers for gaps in TSC FDA-approved medications on state Medicaid formularies and assess the prevalence of prior authorization denials. <ul style="list-style-type: none"> In 2025-2028, work with industry and health policy legislators to advocate for change in access criteria in a minimum of five states. Develop medical necessity letters for those therapies which have the most frequent denials or obtain new FDA-approvals and disseminate them to healthcare professionals. Annually update website to indicate which clinics offer telehealth across state lines. | <p>Short-Term Access/Quality of Care Outcomes <i>Within 5 years:</i></p> <ul style="list-style-type: none"> Reduced barriers to therapeutic treatments and TSC specialists Improved access to comprehensive, high-quality care for those with TSC across the lifespan. Expanded knowledge to inform surveillance and management guidelines for TSC, and updated guidelines disseminated to clinicians. <p>Long-Term Access/Quality of Care Outcomes <i>Within 5 to 10 years:</i></p> <ul style="list-style-type: none"> Widespread awareness and adoption of evidence-based guidelines for TSC surveillance and management among TSC Clinics, COEs and affiliated providers. . | <p>Improve short- and long-term clinical outcomes for individuals affected by TSC through reduced barriers to care and treatments and refinement and dissemination of evidence-based clinical guidelines.</p> |
| | | | <p>Improve quality and comprehensiveness of healthcare:</p> <ul style="list-style-type: none"> Promote access to high-quality, patient-centered clinical care across the lifespan by promoting adherence to evidence-based standards of care at recognized TSC Clinics, COEs and affiliated providers. Integrate behavioral, mental health, social and emotional wellbeing into TSC Clinics, COEs and affiliated providers. | <p>Improve quality and comprehensiveness of healthcare</p> <ul style="list-style-type: none"> Develop a transition plan template based on input from Clinic Committee by 2025 and require its use by 100% of recognized clinics by 2028. In 2024, assess baseline number of TSC Clinics and COEs that have action plans describing TAND service capabilities for both pediatrics and adults. By 2028, double the number of those with action plans relative to baseline. | | |
| | | | <p>Build evidence base to support updates to best practice guidelines for TSC clinical care:</p> | <p>Build evidence base to support updates to best practice guidelines for TSC clinical care:</p> <ul style="list-style-type: none"> Collaborate on initiatives to build evidence to | | |

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| | | | <ul style="list-style-type: none"> Support initiatives to build evidence to address gaps in current surveillance and management guidelines of TSC care (e.g., including reproductive and perinatal health – in collaboration with The LAM Foundation). | <p>address gaps on two understudied aspects of TSC.</p> <ul style="list-style-type: none"> Publish updated consensus guidelines for surveillance and management of TSC for areas previously unaddressed and identified as high priority by the community (e.g., reproductive and perinatal health, SUDEP). | | |
| | | | <p><u>Support and Empower Constituents</u></p> <p>Develop a more collaborative and inclusive volunteer network to mentor the next generation:</p> <ul style="list-style-type: none"> Strengthen our volunteer leadership model to address gaps in resources, support, advocacy and medical access and train our leaders to represent a more inclusive and collaborative community. | <p><u>Support and Empower Constituents</u></p> <p>Develop a more collaborative and inclusive volunteer network to mentor the next generation:</p> <ul style="list-style-type: none"> Fully implement 15 regions by 2024, with key leadership roles represented in each region by December 2028. Develop an intranet platform that facilitates effective communication, training and resource sharing among our volunteer leadership team by 2024 with 90% utilization annually 2025-2028. Develop a benchmark for peer-to-peer support in 2024 and increase documented meetings and interactions by 10% annually in 2025-2028. Recruit and train at least one government advocate in each state by 2028. | <p>Short-Term Support Outcomes <i>Within 5 years:</i></p> <ul style="list-style-type: none"> Empowered volunteer leaders to effectively mentor the next generation. Strengthened volunteer model infrastructure to address gaps in volunteer supports, resources and peer-to-peer engagement. Increased data collection resulting in a 50% improvement to track peer support engagement. Increased access to care for TSC families by collaborating with local TSC Clinics and healthcare providers. | <p>Empower our volunteer leaders to foster an inclusive and collaborative community that is engaged and equipped to provide sustainable support, resources and funding.</p> |
| | | | <p>Strengthen the relationship between healthcare providers and community members at a local and regional level:</p> <ul style="list-style-type: none"> Establish a stronger collaboration with TSC Clinics, healthcare providers and leaders to further support and provide resources to TSC families. Improve TSC community support and resources through cultivating and growing ties with new and current partners. | <p>Strengthen the relationship between healthcare providers and community members at a local and regional level:</p> <ul style="list-style-type: none"> Grow Clinic Ambassador/Medical Liaison program from 20% to 45% of TSC Clinics by 2028. Support and educate 75 TSC families through the IEP and transition processes annually. Increase the number of engagements through the TSC Support Navigators by 10% annually from a baseline of 50 in 2023. Maintain working partnerships with at least 15 national and international organizations/collaboratives (e.g., ARC, CNF, CTF, F/REN, LAM Foundation, ATS-PAR, ELC, Global Genes, ISAN, NORD, RDCRN, TSCi, Mind the Gap, Got Transitions, TANDem). | <p>Long-Term Support Outcomes <i>Within 5 to 10 years:</i></p> <ul style="list-style-type: none"> A 25% increase in representation of underserved communities in our volunteer leadership through recruitment and engagement efforts. | |
| | | | <p>Foster international relationships and development of resources to better serve the worldwide TSC community:</p> <ul style="list-style-type: none"> Grow global expertise as a way to better support the TSC community. | <p>Foster international relationships and development of resources to better serve the worldwide TSC community:</p> <ul style="list-style-type: none"> Grow global clinics from 13 to 25 by 2028. Work with TSCi to increase the number of countries participating in TSC clinical trials by 25% by 2028. Foster partnerships in 10 low- or middle-income countries with no association representation by 2028. | | |